

Friedreich's Ataxia

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Friedreich's Ataxia (FRDA) is an inherited (genetic) progressive disorder of the nervous system that affects balance, coordination, movement, and sensation." Ataxia" means a loss of muscle coordination, which is usually the earliest and most prominent characteristic of the disease.

Alternate Names

Friedreich's Ataxia is also known as familial ataxia, Friedreich's disease, Friedreich's tabes, hereditary ataxia-Friedreich's type, Spinal ataxia – hereditofamilial or spinocerebellar ataxia.

Causes, incidence and risk factors

Friedreich's Ataxia is caused by an abnormality in a gene, called X25. Genes are sets of instructions that tell the cells, containing chromosomes, how to build the proteins that enable the cells to carry out their various functions. These functions determine a person's physical characteristics, from the color of the hair and eyes to the organization of the nervous system.

Friedreich's Ataxia occurs when there is a lack of the protein frataxin (for which X25 provides the code) in the tissues. The lack of protein causes the nerve cells within the tissues of the spinal cord and its brain connections, the heart and pancreas to degenerate. When degeneration happens the nerve signals to the muscles are reduced.

Friedreich's Ataxia develops when a person inherits the defective gene from both parents. This is a recessive inheritance pattern. If only one parent contributes a defective gene, the child becomes a "carrier" of Friedreich's Ataxia but never develops the disorder. It is estimated that 1 person in every 100 people in the general population is a carrier of the Friedreich's gene defect and 1 in every 40,000 is affected with Friedreich's Ataxia. A child of parents who are both carriers has a 25 percent chance of inheriting the disease.

Symptoms

The symptoms usually begin in childhood or youth (age 5 through 25) as a result of the deterioration in areas of the brain controlling muscle coordination, the spinal cord and nerves. Symptoms may include:

- Clumsiness - Progressive weakness of the legs which may appear as a staggering or lurching gait
- Reduced muscle coordination
- Trembling when standing still
- Partial loss of sense of touch or sensitivity to pain and temperature
- Arms and legs may become weak or numb
- Paralysis of the lower limbs



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- Impaired speech
- Impaired swallowing
- Spine may begin to curve to one side (scoliosis)
- Feet may become rigid and deformed
- Vision problems
- Hearing problems
- Diabetes develops
- Heart muscles may be impaired (cardiomyopathy)
- Tremors: use of propranolol
- Muscle spasms: use of dantrolene sodium
- Curvature of spine/foot deformities: orthopedic surgery or braces
- Vision problems: corrective devices such as glasses and contact lens, surgery or medication
- Hearing problems: hearing aids, surgery or medication
- Muscle function: the use of physical therapy
- Cardiomyopathy: diuretic and antiarrhythmic drugs or heart transplant

Diagnosis

The diagnosis is based on a person's medical history, family history, physical exam and a complete neurological evaluation, which includes an electromyography (EMG). An EMG is a test in which the electrical activity in muscle is analyzed after being amplified, displayed and recorded. A nerve conduction velocity test is also used sometimes. Small needles are inserted below the skin to determine the strength and speed of electrical impulses traveling along the nerves. In FRDA, the strength of these signals is abnormal.

Genetic testing has become the most accurate way to diagnose FRDA since the frataxin gene has been identified. The flaw in the frataxin gene causes a "triplet repeat" in which the section of DNA is repeated over and over. Genetic testing examines the DNA from a blood sample to look for the triplet repeats associated with FRDA.

Treatment

Since FRDA is a degenerative disease of the nervous system there is no specific treatment. However, many of the symptoms associated with FRDA can be treated or controlled. Treatment methods for symptoms are listed below:

- Diabetes: use of insulin

More information regarding Friedreich's Ataxia may be located on the following web sites:

Friedreich's Ataxia Parent's Group
www.fortnet.org/fapg

National Ataxia Foundation **www.ataxia.org**

International Network of Ataxia Friends
www.internaf.org

Arkansas Chapter of the National Ataxia Foundation:

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References

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Muscular Dystrophy Association. (2003). Facts about Friedreich's Ataxia [Electronic Database]. MDA (Producer). MDA Publications (Distributor).

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